

NOTES

LOWER RESPIRATORY TRACT CONGENITAL MALFORMATIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Structural anomalies of lung during embryonic development

DIAGNOSIS

Prenatal ultrasound, radiography, CT scan, MRI

SIGNS & SYMPTOMS

- Asymptomatic/respiratory distress

TREATMENT

- Surgical resection, respiratory support

CONGENITAL PULMONARY AIRWAY MALFORMATION

osms.it/cpam

PATHOLOGY & CAUSES

- Congenital pulmonary airway malformation (CPAM)
- AKA congenital cystic adenomatoid disorder → part of/entire lung lobe replaced by non-functional cysts
- Result of abnormalities in branching morphogenesis of lung
- Rare in general; most common congenital lung malformation

TYPES

- CPAM Type 0: rare; multiple small cysts composed of cartilage, mucus cells; lethal anomaly
- CPAM Type 1: most common; large (> 2cm/0.78in) multiloculated cysts; good prognosis
- CPAM Type 2: small uniform cysts;

associated with other congenital malformations (e.g. esophageal atresia with esophageal fistula); poor prognosis

- CPAM Type 3: large; not true cysts; communicates with surrounding parenchyma; “adenomatoid” type; more common in individuals who are biologically male
- CPAM Type 4: rare

CAUSES

- Possibly, *in utero* airway obstruction/atresia; definitive cause unknown

RISK FACTORS

- Occurs sporadically
- Not related to maternal factors
- No genetic predisposition except Type 4 (associated with familial pleuropulmonary blastoma syndrome)

VISIT...

LANZAROTE
Caliente.COM

COMPLICATIONS

- Pulmonary hypoplasia
- Mediastinal shift, putting pressure against heart
- Respiratory infections
- Rapid growth → venous outflow obstruction, cardiac failure, hydrops fetalis, death
- Longstanding CPAMs → cancer

SIGNS & SYMPTOMS

- 75% of individuals: asymptomatic
- 25% of individuals: cyanosis, pneumothorax, respiratory distress, tachypnea, intercostal retractions, grunting
- Hyperresonance on percussion, diminished vesicular murmur, asymmetrical thorax

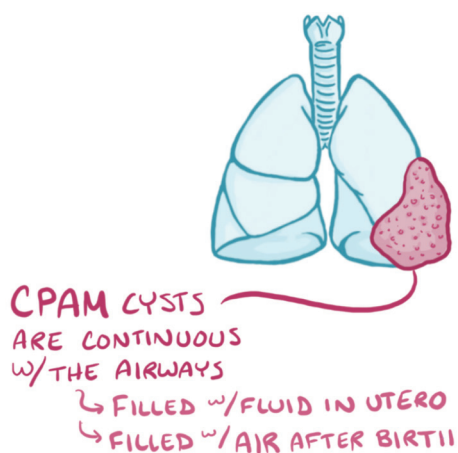


Figure 124.1 Illustration depicting continuous pulmonary airway malformation.

DIAGNOSIS

- Definitive diagnosis usually not possible without surgical resection, histopathological evaluation

DIAGNOSTIC IMAGING

Prenatal ultrasound

- Echogenic mass appearing in the chest, displacement of heart, flat/everted

diaphragm, absence of visible lung tissue

MRI/CT scan

- Delineate pathology

TREATMENT

SURGERY

- Minimally invasive surgical resection (thoracoscopy)
- **Large cysts:** *in utero* placement of Harrison thoracoamniotic shunt
- **Rare:** fetal surgery *in utero*; surgical delivery, *ex utero* intrapartum treatment (EXIT) procedure

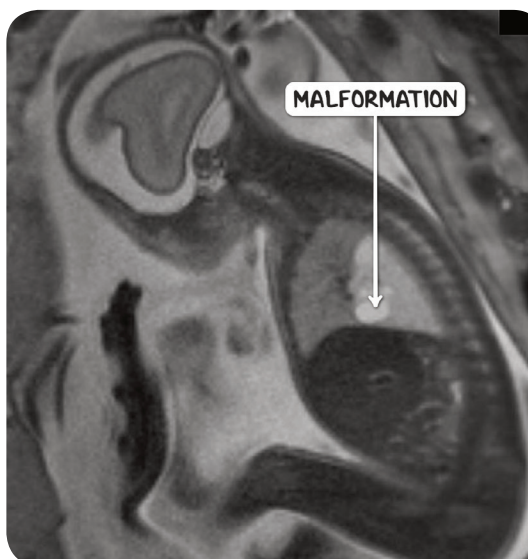


Figure 124.2 A fetal MRI demonstrating a congenital pulmonary airway malformation.

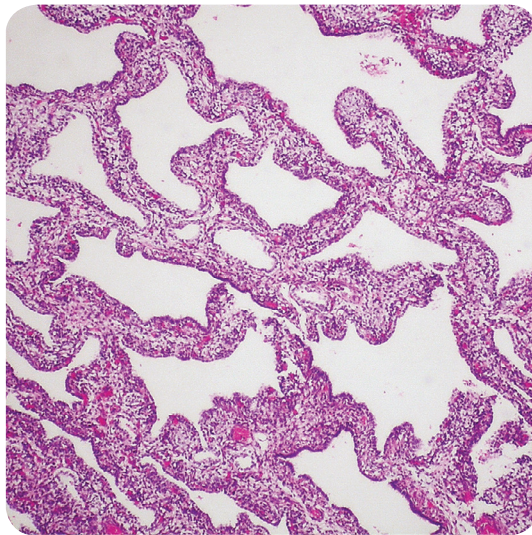


Figure 124.3 The histological appearance of a congenital cystic type II malformation. There are multiple small cystic spaces lined by immature respiratory tissue.

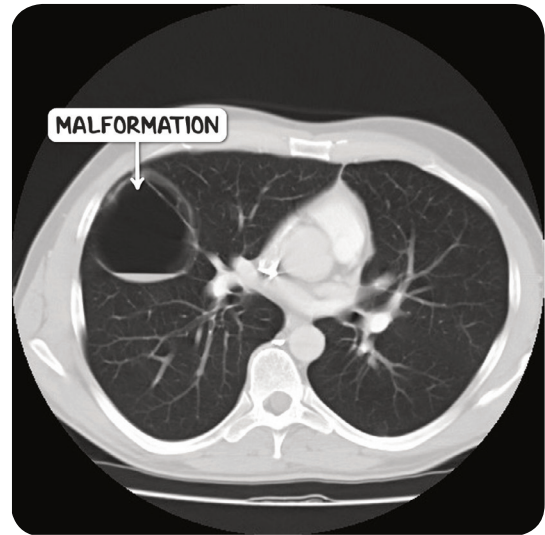


Figure 124.4 A CT scan of the chest demonstrating a pulmonary congenital cystic adenomatoid malformation, Type I, presenting as a single cyst of middle lobe in an adult.

PULMONARY HYPOPLASIA

osms.it/pulmonary_hypoplasia

PATHOLOGY & CAUSES

- Underdevelopment of lungs → **low number/size of bronchopulmonary segments/alveoli**
- Typically occurs prior to/after pseudoglandular stage (6–16 weeks gestation)

TYPES

- Primary
 - Idiopathic, not associated with maternal/fetal abnormalities; rare
- Secondary
 - Due to **fetal abnormalities** disrupting lung development
 - Associated with **bilateral renal agenesis, congenital diaphragmatic hernia, congenital cystic adenomatoid malformation, fetal hydronephrosis,**

caudal regression syndrome, mediastinal tumor, dextrocardia, sacrococcygeal teratoma

- Bilateral renal agenesis → oligohydramnios → decreased lung expansion, decreased mechanical stretching → decreased growth factors lung synthesis → pulmonary hypoplasia

RISK FACTORS

- **Decreased amniotic fluid:** severe oligohydramnios, mid-trimester rupture of membranes
- **Disruption of signaling pathways involved in growth:** sonic hedgehog (SHH) signaling pathway
- **Aberrant expression growth factors:** vascular endothelial growth factor (VEGF), epidermal growth factor (EGF), fibroblast growth factor (FGF)
- Early delivery

COMPLICATIONS

- **Respiratory distress**, chronic respiratory failure, bronchopulmonary dysplasia, pneumothorax, secondary scoliosis, impaired cardiac function
- Survival depends on degree of hypoplasia, cause of restricted growth

SIGNS & SYMPTOMS

- **Prenatal**: poor fetal movement, amniotic fluid leakage, oligohydramnios
- **Postnatal**: asymptomatic/severe respiratory distress, apnea, cyanosis
- Small, bell-shaped chest; heart displacement; decreased/absent breath sounds

DIAGNOSIS

DIAGNOSTIC IMAGING

3D ultrasound

- Total lung volume measurement

Doppler ultrasound/magnetic resonance angiography

- Shows lack of blood supply

CT scan/MRI

- Shows loss of lung volume

Radiography

LAB RESULTS

- Renal function (serum creatinine, blood urea, electrolyte levels)
 - Oligohydramnios

OTHER DIAGNOSTICS

- Lung weight, lung weight to body weight ratio, mean radial alveolar count (RAC), lung DNA

TREATMENT

OTHER INTERVENTIONS

- **Amnioinfusion**: instilling isotonic fluid into amniotic cavity
- **Amniopatch**: intra-amniotic injection of platelets, cryoprecipitate → seal amniotic fluid leak
- Treatment of underlying condition; respiratory support; in severe cases, fetal surgery



Figure 124.5 A chest X-ray demonstrating a volume defect of the right thoracic cavity caused by pulmonary hypoplasia.